SLIDE - What is a “Rare Disease”?

So, what exactly IS a rare disease?

In the US,

a rare disease is categorized as a disease that affects fewer than 200,000 people.

According to the National Institutes of Health, There are more than 6,800 defined rare diseases,

which on the whole,

affect 25-30 million Americans.

This works out to be nearly 1 in 10 of us.

Doesn’t sound so “rare” does it?

Because of the lack of knowledge and lack of medical information on these diseases, there is often high numbers of misdiagnosis and delayed diagnosis. Leaving families feeling helpless and frustrated throughout the process.

SLIDE - Types of Rare Diseases…

A significant portion (believed to be around 80%) of rare diseases are genetic in nature, called “rare genetic diseases” creative I know,

this means they are cause by some type of mutation of a specific gene.

Many times, these mutations can be passed from one generation to the next,

explaining why there are family histories of certain diseases.

However, genes do not make up all reasons for rare diseases and in some other cases,

There are factors such as

environment,

diet,

and exposure to toxins or chemicals,

Rare diseases also span all areas of the body,

neurological,

muscular,

skeletal,

body organs and systems.

All can be affected.

SLIDE - Some Examples of Rare Diseases…

Some examples of rare diseases, we can see here:

 Primary Ciliary Dyskinesia (PCD)

Which is the disease that Angelina has, affects the cilia. We’ll get some details on PCD in a minute.

* Niemann-Pick Type C (NPC)

That just this week, had a page long article in the Wall Street Journal’s Health and Wellness section with the story of the race to save two twin 9 year old girls diagnosed with NPC. I encourage you to read it - web

* Hereditary breast and ovarian cancers –

which have been in the limelight lately

as women are now able to have genetic testing performed- Angelina Jolie

that can show specific mutations in the BRCA1 And BRCA2 genes,

and helps them determine a proactive care plan

as opposed to waiting for a cancer diagnosis.

* Cystic Fibrosis
* Muscular Dystrophy

And many, many more.

SLIDE - PCD or Primary Ciliary Dyskinesia…

Our organization, will serve the entire rare disease community, however, we do have a focus on PCD since that is our personal connection to that community.

Primary Ciliary Dyskinesia is a rare genetic disease believed to affect about 25,000 people in the US, with only around 400 having an official diagnosis.

PCD is inherited in an

autosomal recessive manner.

This means that both parents must carry the mutated gene in order for the children to be affected. Carriers of the disease are asymptomatic and would not have any knowledge that they were carriers.

**SLIDE** - There are 17 genes known to be associated with PCD and among those, mutations in DNAH5 account for about 15-20% of all cases.

**SLIDE -** Each sibling of an affected PCD individual has a 25% chance of having PCD as well, a 50% chance of being a carrier, and a 25% chance of being completely unaffected and a non-carrier.

If the disease is known in the family, there CAN be carrier testing for relatives, and prenatal testing for women who have a known increased risk.

**SLIDE** - PCD is a disease that affects the cilia in your body, or the little hair like structures, that most of us know to have in our lungs, which are actually in your sinuses, ears, reproductive organs, and ventricles of the brain as well.

In a healthy individual, cilia moves or “beats” back and forth together, looking much like seaweed in the ocean, and helps keep lungs, sinuses, and ears free of organisms and debris that can cause infections.

Individuals with PCD have cilia that either do not beat at all, or beat in abnormal patterns. This causes an inability to clear airways of mucus. When this happens, the respiratory tract, lungs, and airways become very inflamed and a perfect breeding ground for bacteria as the mucus sits there.

Consequently, PCDers have chronic infections in their lungs and sinuses, bouts of pneumonia, and can also develop a stretching and weakening of the bronchial tubes called bronchiectasis.

For reasons not quite known for sure, PCDers also tend to have stomach issues as well as headaches. Some explanations for these are medication side effects, and the presence of constant sinus pressure and draining.

**SLIDE** - In the womb, cilia also play a role in determining organ placement or “situs” in the body. Because of this, about 50% of PCDers also have a condition called situs inversus. This is where your organs are in an exact mirror image of where they are normally. It does not affect the function of the organs, just placement.

Individuals who have PCD as well as situs inversus are refered to most commonly as having Kartagener's Syndrome, named after the Dr. who linked situs inversus with sinusitis and bronchiectasis.

PCD also often affects fertility - Most males with PCD have very low sperm motility, making them infertile, but not sterile.

Women with PCD may experience some infertility, with a higher risk for miscarriage or ectopic pregnancy. However, some conceive with no problems at all.

In very rare instances, PCD may be associated with hydrocephalus, a condition in which excess fluid in the ventricles of the brain causes them to be enlarged.

SLIDE - Diagnosis…

So, how do you know that you have PCD…

There are currently only two officially recognized ways to make the diagnosis of PCD:

A biopsy that evaluates the internal components of the cilia,

and genetic testing.

Ultimately, genetic testing will be the best, most reliable method for making the diagnosis, but currently available genetic testing does not pick up all known mutations for PCD, and therefore is not widely performed.

For this reason ciliary biopsy,

performed at a center trained to collect, process and analyze cilia for PCD

is still the ‘gold standard’ for diagnosis.

After a biopsy is taken, results can be expected in about 6-8 weeks.

SLIDE - Treatments…

There are no standard approved treatment plans for PCD, however, there are guidelines that are recommended by PCD experts in the field.

Medications:

There are some main drugs that many PCDers use…those are mucus thinners, airway relaxers, and antibiotics and steroids.

These are administered in various ways, but many are inhailed through a nebulizer – a machine that vaporizes liquid so it can be breathed into the lungs, or inhailer.

Antibiotics treat the bacterial infections, and streoids battle the inflammation in the airways and lungs. Most PCDers do extended rounds of antibiotics at maintenance levels to keep infection at bay, cycling on and off throughout the year.

Airway Clearance Therapy or (ACT)

Are therapies or activities designed to help clear mucus from the airways.

ACT can be done in a few ways.

Hand cupping and vest therapy are the two main forms used in PCD,

primarily because there is a need for daily, clearance.

**SLIDE -** “The Vest” as it’s so fondly called is ACT equipment that helps to shake loose mucus that is present in smaller airways so that it can be moved to larger airways and more easily expelled –

here is Angelina wearing hers, she wanted camo, no pink for this little warrior!

It also costs $18,000 and comes with a title – like your car

She got her vest in April, right before my birthday, and on my birthday, she called me up and sang me a vibrating version of happy birthday

The most important thing about airway clearance therapy is that you are #1 doing it consistently, every day and #2 that you are coughing out the mucus that is being moved.

There is little benefit to moving mucus and then letting it sit in the lungs, PCDers need to cough…a lot…

There are also a few procedures that PCDers have to go through pretty routinely. These include bronchoscopy where mucus can be cleaned out of the lungs using tubes, and collected and analyzed. Mucus cultures, to check for infection levels. Nasal rinses and nasal cleanout procedures, as well as ear tubes and hearing aids.

In addition to these medical treatments, it is very important that individuals with PCD lead very active lives. They should exercise and play as much as possible because that keeps lungs healthy and everything moving.

SLIDE - Our Story…

So now that you have a little knowledge as to what rare diseases and PCD are,

let’s talk about the story behind One Breath, One Hope.

**SLIDE -** A little over 6 years ago, a little girl named Angelina was born to Sara and Mel Manwaring,

my very, very good friends.

They had already had one little girl

about 5 seconds after they got married

so Angelina was their second.

**SLIDE -** Mikayla, her big sister was very excited

and I found out that I was going to be able to have the great honor of calling Angelina my godchild.

Angelina had no signs of disease when she was born

but soon battled some health problems that were not so normal.

She had many ear infections,

was diagnosed with RSV only a short time after birth,

and started getting pneumonia before age 2.

**SLIDE -** As the next couple years went on,

visits to the doctor and emergency room were increasingly common to help with breathing issues,

recurring pneumonia,

and flu like symptoms.

Angelina,

along with Sara and Mel,

went through countless sleepless nights filled with violent coughing spells and vomiting.

With very few answers and even less relief from symptoms,

they experienced months of hopelessness and confusion as failed treatments and unclear test results continued to give no answers as to what might be causing the issues.

It was a pretty dark time for them all.

Finally, after all other testing had been exhausted, and no clear reason for the continued illness had surfaced,

their pulmonologist performed a biopsy of Angelina’s lung cilia to test for a rare genetic disease called PCD.

It was a long 6 weeks to receive the news,

and when a positive result came back, it was a mix of fear of what this now meant,

and relief to at long last have a definitive diagnosis,

and hopefully some kind of treatment plan.

As we all poured through online resources to read about this extremely rare disease

we began to feel overwhelmed.

Angelina was put on a whole host of medications, a vest was ordered for her, and the trial and error of treatment that they had been going through, began again.

Now, several months after the diagnosis, things are beginning to look up.

Angelina is being seen by the completely amazing Doctors and Staff at Boston Children’s Hospital,

which has a specialized PCD center on the campus.

Together with the team of Doctors and nurses, we are learning more and more about her specific case.

She has gone through many long drives, blood draws, procedures, and prodding and through it all, she still has kept her fighting personality and brilliant smile.

Now, all of that is a lot as it is…however, there’s more…

In the midst of all of Angelina’s sickness, Mel lost his job just as Sara was getting ready to give birth to Bubba, their third child, and finally a boy!

**SLIDE -** When the baby was born they found out that he was ALSO affected with an extremely rare genetic disease called galactacemia and the black cloud continued. Fortunately, he is ok now, as I’m sure you’ve seen him running around with the other kids here, but the pressure on the family was huge.

Eventually, after no luck in finding any type of job here in CT, Sara and Mel were forced to declare bankruptcy, sell their home, and move out of state to a home in Georgia that Sara’s parents had close to family there. That decision in itself was hell to make.

Being away from their doctors,

their friends,

most of their family

and the lives they had known

was a huge strain.

Then, thankfully about a year later,

a new job opportunity came up and they were able to move back home to Connecticut

and to all of us who missed them so much.

Although,

I will admit,

I flew down to make the drive back with Sara and the kids while Mel drove the moving truck…

and a little distance after being in a car with 3 kids all under the age of 8 for 2 days was not a bad thing!

This whole experience has been scary, and frustrating and exhausting for their family.

**SLIDE -** However, for those of us who were forced to sit and watch, helpless to do anything for them, it was no better.

We held their hands and sometimes their cocktails, as we tried to just be there. It was about all we COULD do.

But as time went on and we learned more and more about the struggles facing the PCD community and rare disease community as a whole, we decided that there was something good that had to come of all this bad.

We wanted to find ways to help Angelina and her family, but also the numerous other families that we knew had to be going through the same thing.

We had to act.

What came of that decision to do SOMETHING,

is One Breath, One Hope.

**SLIDE -** We began this non-profit with the hope of helping people deal with the struggles that come with rare disease diagnoses.

Our mission is to provide financial relief,

where we can,

to help with things like travel to reach specialized doctors,

medical expenses that aren’t covered by insurance,

technology purchases that will help kids like Angelina to keeping current in their school work,

and anything else that might give these individuals and families a moment of hope and a breath of relief on this long road they are travelling.

We are also partnering with research organizations like The PCD Foundation to advocate for rare disease research,

and utilizing social media outlets like Facebook to try connect people to each other and encourage a real community to form,

which is so important to alleviating the feeling that they are going through this alone.

To the best of our knowledge there are no other organizations like us. We are unique in that we are not raising funds for research (which is hugely important), but rather raising funds to help the people.

If we can give one family every year, a good day, then we will have accomplished something great.

We will be publishing a quarterly newsletter,

starting in January,

I hope all of you have added your names and addresses to our mailing list

because your ticket this evening has covered your membership (that costs $20) to OBOH and I consider you all part of the OBOH family now.

In that newsletter we will have stories of the people we help, like Angelina,

as well as any developments we have heard of in the PCD and rare disease community.

We would also like to include any topics that you would like to hear about.

On the back table, there are some notecards and pens, if you have a question or topic you would like to see covered in one of our newsletters, please write it down and put it in the bowl.

We also have a few pamphlets about OBOH and about PCD if you would like to take those home with you and please take some time to go to our website and poke around [www.onebreathonehope.org](http://www.onebreathonehope.org)

Education and awareness are our best allies in this battle.

**SLIDE -** And with the formal presentation being over, I’d like to ask if anyone has any questions tonight that they would like to ask?

Q&A